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Statistical Weights of Mixed DNA Profiles

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Statistical weights of mixed DNA profiles

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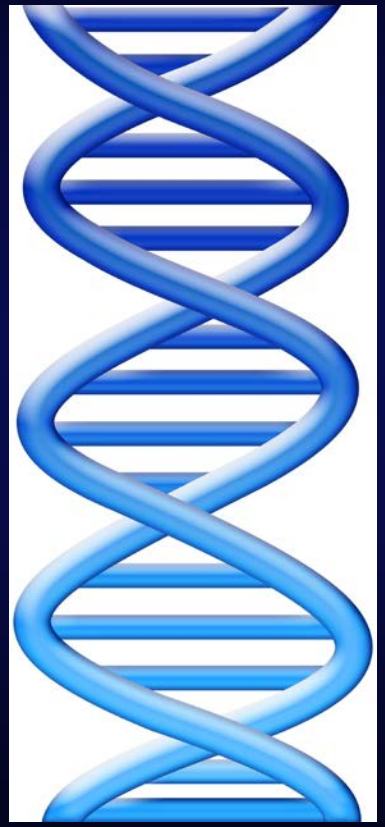
Forensic DNA Profiling Video Series

Forensic Bioinformatics
(www.bioforensics.com)

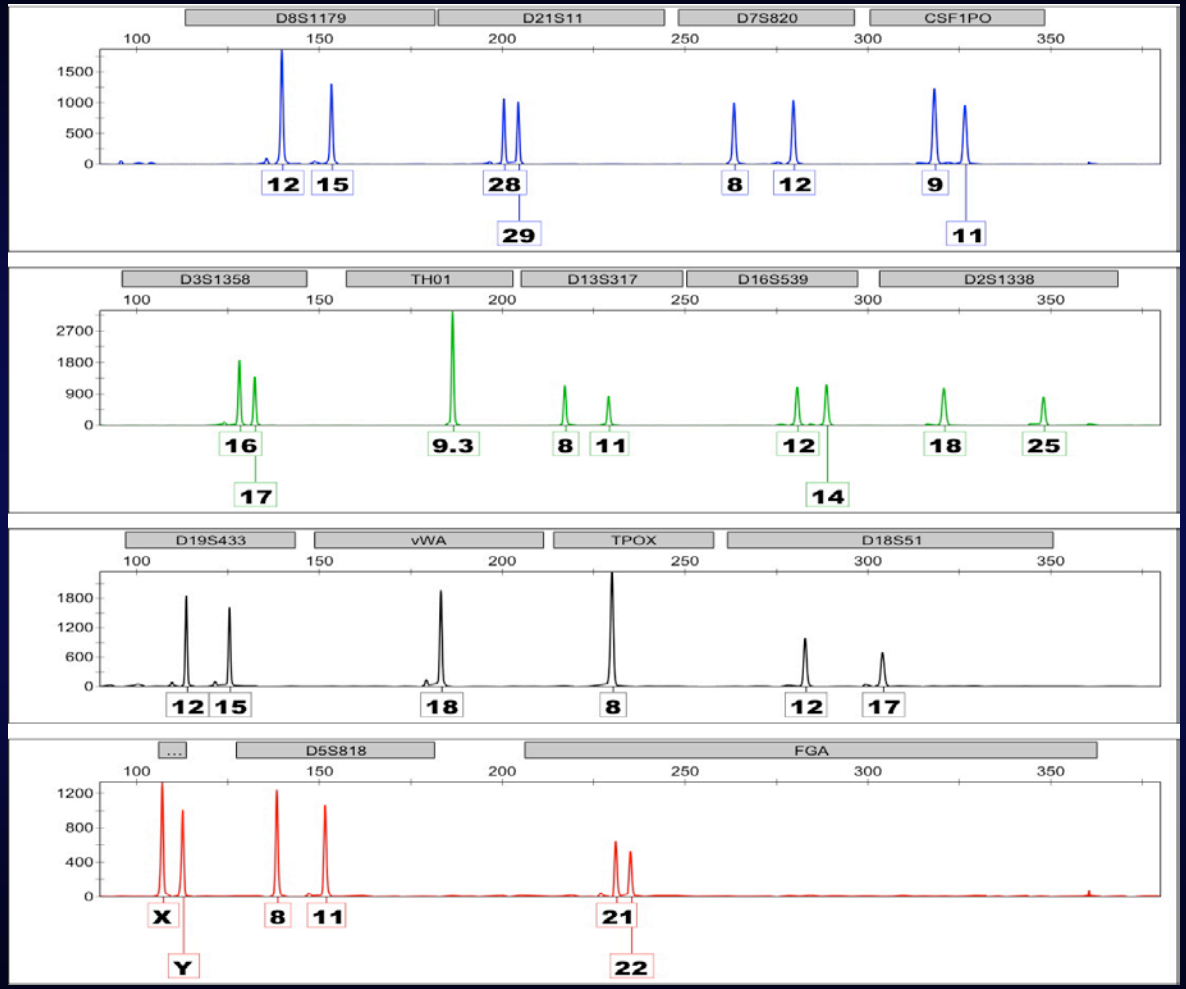
DNA statistics



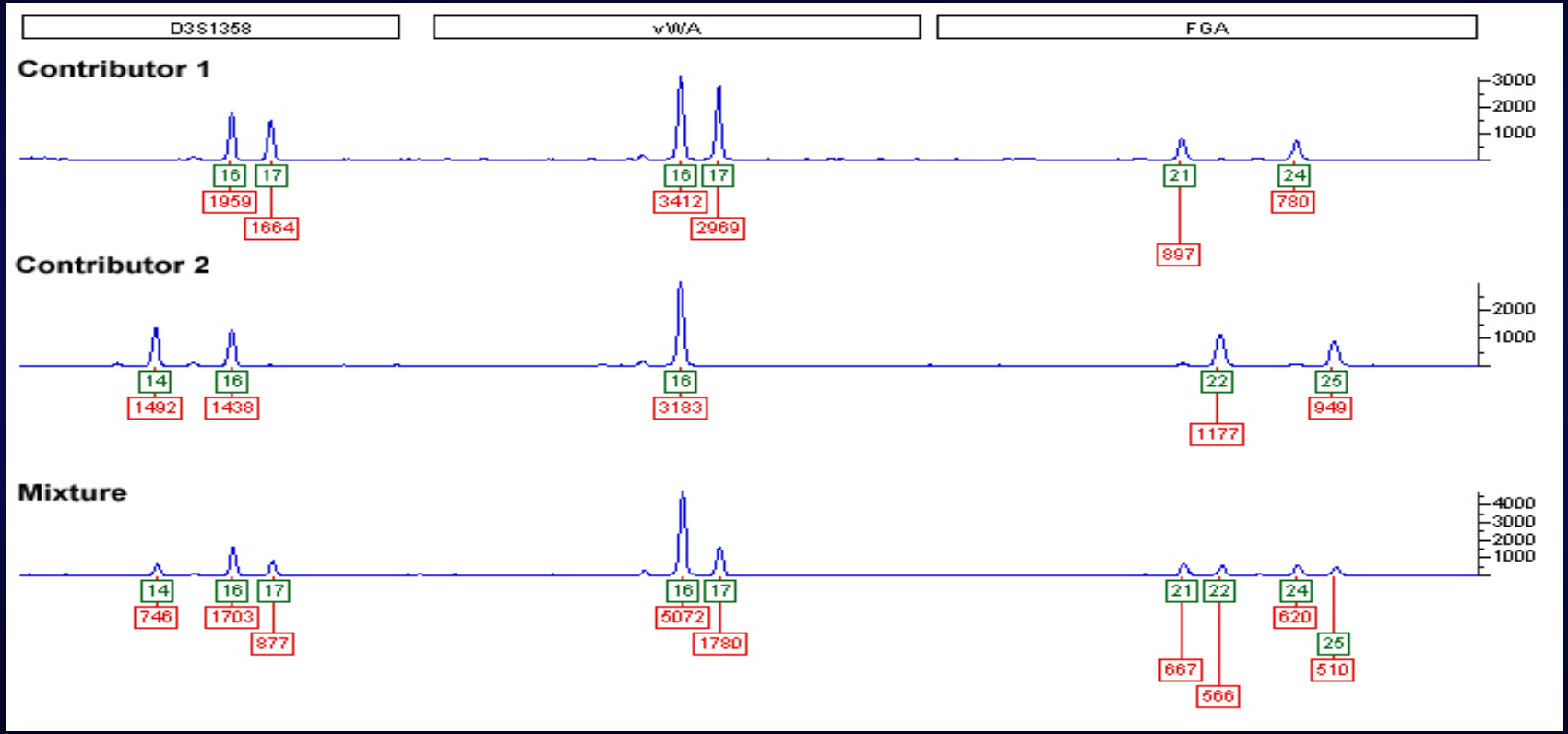
- Single source samples can be easy to interpret
- Several factors can make statistics less impressive
  - Mixtures
  - Incomplete information
  - Relatives

DNA profile

LOCUS	ALLELES
D8S1179	12, 15
D21S11	28, 29
D7S820	8, 12
CSF1PO	9, 11
D3S1358	16, 17
TH01	9.3
D13S317	8, 11
D16S539	12, 14
D2S1338	18, 25
D19S433	12, 15
vWA	18
TPOX	8
D18S51	12, 17
AMEL	X, Y
D5S818	8, 11
FGA	21, 22



Mixed DNA samples



How many contributors to a mixture? If analysts can discard a locus?

Maximum # of alleles observed in a 3-person mixture	# of occurrences	Percent of cases
2	0	0.00
	3,398	0.00
3	78	0.00
	26,788,540	18.28
4	4,967,034	3.39
	112,469,398	76.75
5	93,037,010	63.49
	7,274,823	4.96
6	48,532,037	33.12

There are 146,536,159 possible different 3-person mixtures of the 959 individuals in the FB I database (Paoletti et al., November 2005 *JFS*).

How many contributors to a mixture? If analysts can discard a locus?

Maximum # of alleles observed in a 3-person mixture	# of occurrences	Percent of cases
2	0	0.00
	8,151	0.02
3	310	0.00
	11,526,219	25.53
4	2,498,139	5.53
	32,078,976	71.07
5	29,938,777	66.32
	1,526,550	3.38
6	12,702,670	28.14

There are 45,139,896 possible different 3-person mixtures of the 648 individuals in the MN BCI database (genotyped at only 12 loci).

How many contributors to a mixture?

Maximum # of alleles observed in a 4-person mixture	# of occurrences	Percent of cases
4	13,480	0.02
5	8,596,320	15.03
6	35,068,040	61.30
7	12,637,101	22.09
8	896,435	1.57

There are 57,211,376 possible different 4-way mixtures of the 194 individuals in the FB I Caucasian database (Paoletti et al., November 2005 *JFS*). (35,022,142,001 4-person mixtures with 959 individuals.)

What weight should be given to DNA evidence?

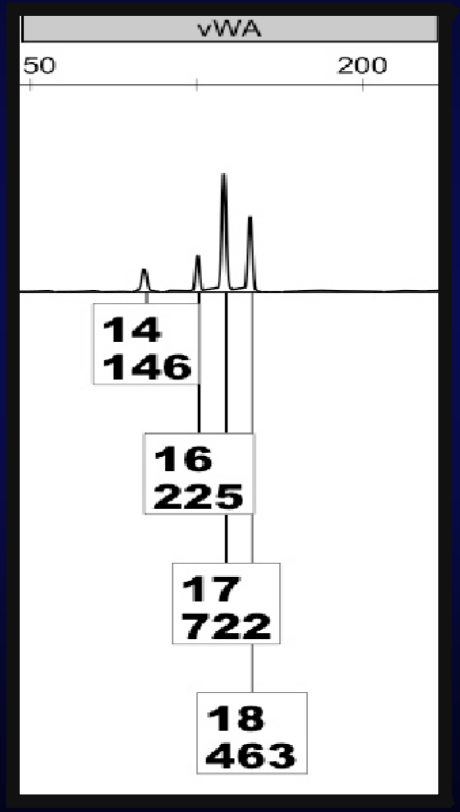
Statistics do not lie.

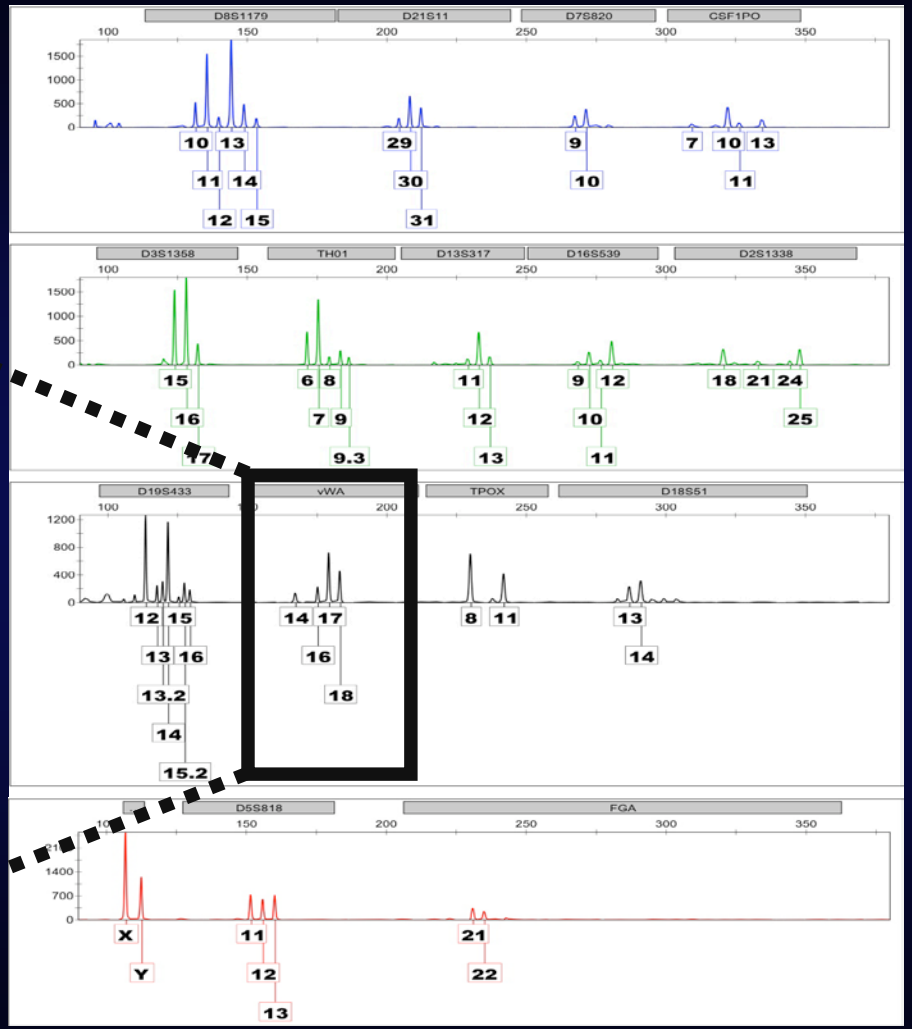
But, you have to pay close attention to the questions they are addressing.

What is the chance that a randomly chosen, unrelated individual from a given population would have the same DNA profile observed in a sample?

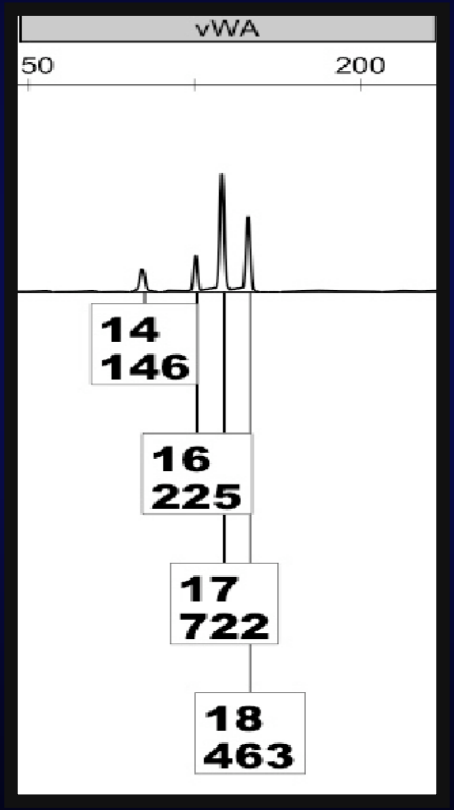
Mixture statistics: Combined Probability of Inclusion (CPI)

Mixture statistics





Combined Probability of Inclusion



- Probability that a randomly chosen, unrelated person could be included as a possible contributor to a mixed profile
- For a mixed profile with the alleles 14, 16, 17, 18; contributors could have any of 10 genotypes:

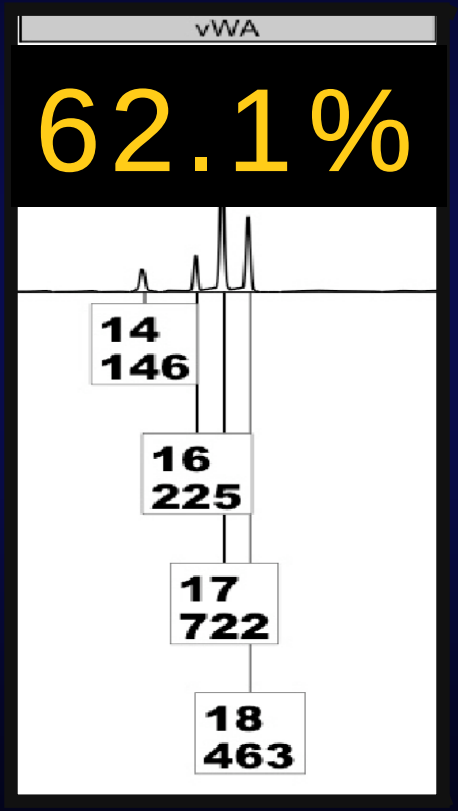
14, 14	14, 16	14, 17	14, 18
	16, 16	16, 17	16, 18
		17, 17	17, 18
			18, 18

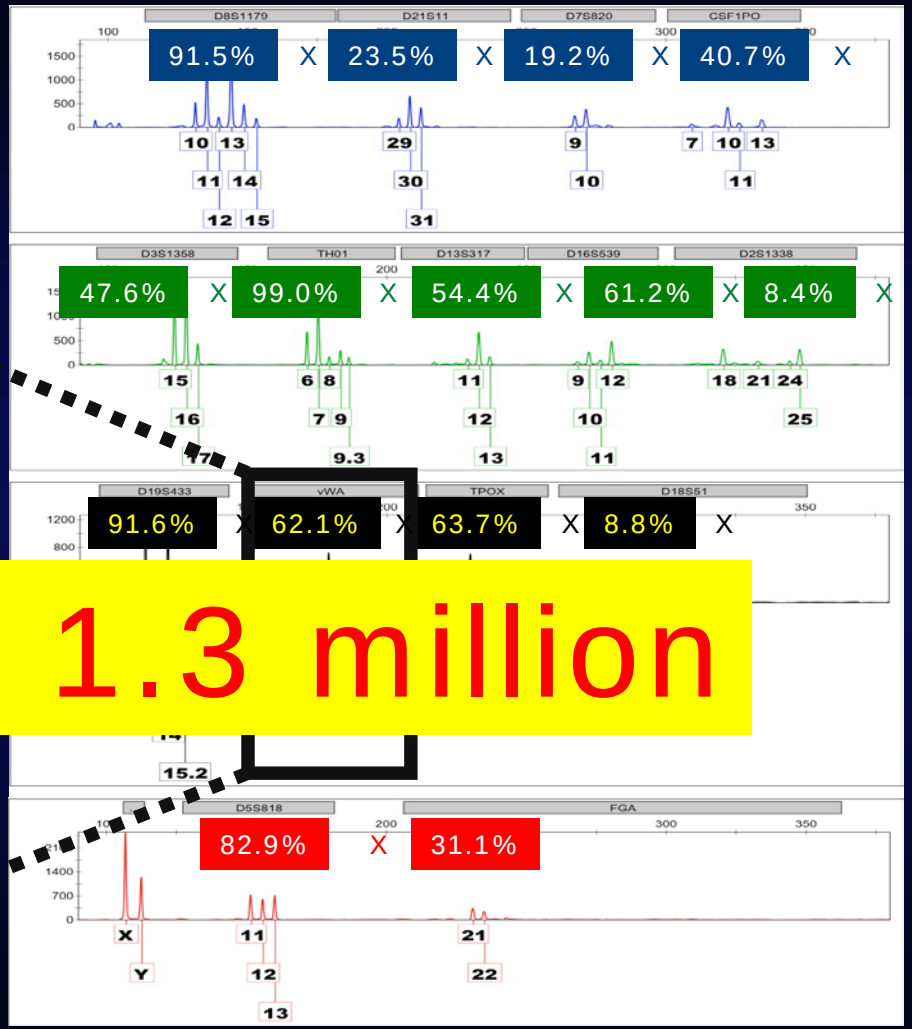
Probability works out as:

$CPI = (p_{[14]} + p_{[16]} + p_{[17]} + p_{[18]})^2$

$(0.102 + 0.202 + 0.263 + 0.222)^2 = \mathbf{0.621}$

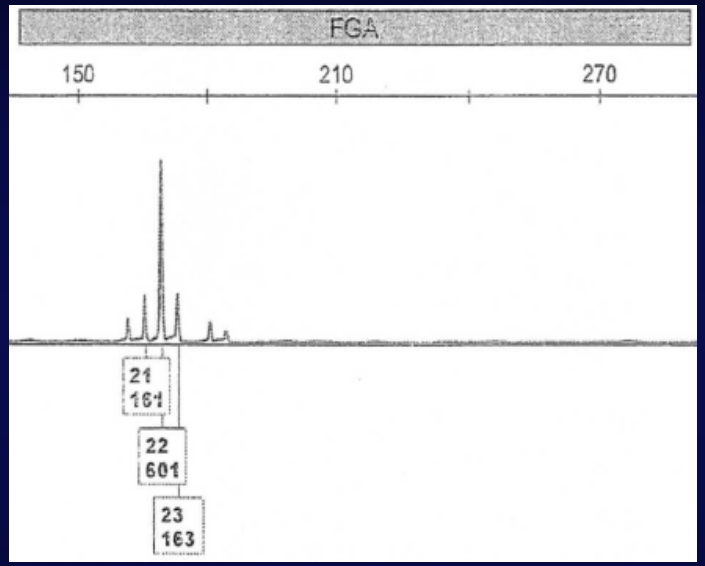
Mixture statistics





1 in 1.3 million

Combined Probability of Inclusion



- What is the chance that a randomly chosen, unrelated person could be included as a possible contributor to a mixed profile?
- For a mixed profile with the alleles 21, 22, 23; contributors could have any of 6 genotypes:

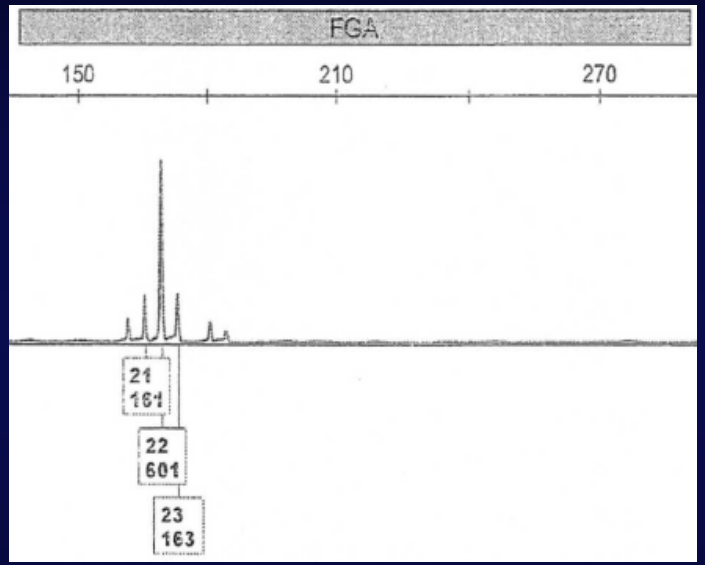
21, 21 21, 22 21, 23

22, 22 22, 23

23, 23

Probability works out as:

Combined Probability of Inclusion



- What is the chance that a randomly chosen, unrelated person could be included as a possible contributor to a mixed profile?
- When allelic drop out can be invoked, CPI no longer applies

$$CPI = (p_{[21]} + p_{[22]} + p_{[23]})^2$$

What statistic applies to an individual who is 22, 25?

What weight should be given to DNA evidence?

Statistics do not lie.

But, you have to pay close attention to the questions they are addressing.

What is the chance that a randomly chosen, unrelated individual from a given population would have the same DNA profile observed in a sample?

Victim: N/A
Suspect: Brendacia Xavier and Franklin Xavier

CERTIFICATE OF ANALYSIS

The following evidence was received by this laboratory on June 10, 2008 by FedEx International delivery:

Agency #	DLI #	Description
1	08-01097	Swabs from Rossi revolver s/n XC71548
4	08-01100	Black holster
2	08-01098	Reference standard from Franklin Xavier
3	08-01099	Reference standard from Brendacia Xavier

RESULTS/CONCLUSION

Samples from items 1, 2 and 3 were extracted for deoxyribonucleic acid (DNA). Those samples with sufficient amounts of DNA were amplified by polymerase chain reaction (PCR) and analyzed using the Applied Biosystems Identifiler and/or MiniFiler profiling system. The tested loci and DNA results are outlined in the attached report table.

LOCUS/ITEM	1 (08-01097) Swabs from revolver-Rossi s/n XC71548	2 (08-01098) Standard from Franklin Xavier	3 (08-01099) Standard from Brendacia Xavier
D8S1179	NT	12,12	14,14
D21S11	28,30	28,30	27,31.2
D7S820	9,10,11	10,11	9,10
CSF1PO	7,8,10,11,12	7,11	8,12
D3S1358	NT	16,17	16,16
TH01	NT	7,8	7,8
D13S317	11,12	11,12	8,12
D16S539	8,9,11,14	8,8	9,11
D2S1338	16,19,20,22,25	16,22	19,20
D19S433	NT	13,13	14,15
vWA	NT	15,17	17,18
TPOX	NT	9,12	8,8
D18S51	12,13,14,16,18	16,18	12,14
Amelogenin	X,Y	X,Y	X,X
D5S818	NT	13,13	11,12
FGA	21,22,23	22,25	21,21

NT- not tested

The testing lab's conclusions

1 08-01097 Swabs from Rossi revolver s/n XC71548

Blood was indicated on the swabs.

A DNA profile was obtained from this item and indicates a mixture of at least three individuals. Franklin Xavier (item 2) cannot be excluded as a possible contributor to this mixed DNA profile. The probability of randomly selecting an unrelated individual matching this mixed DNA profile is approximately 1 in 1,600 individuals. Brendacia Xavier (item 3) cannot be excluded as a possible contributor to this mixed DNA profile. The probability of randomly selecting an unrelated individual matching this mixed DNA profile is approximately 1 in 600 individuals.

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TH01	NT	7,8	7,8
D13S317	11,12	11,12	8,12
D16S539	8,9,11,14	8,8	9,11
D2S1338	16,19,20,22,25	16,22	19,20
D19S433	NT	13,13	14,15
vWA	NT	15,17	17,18
TPOX	NT	9,12	8,8
D18S51	12,13,14,16,18	16,18	12,14
Amelogenin	X,Y	X,Y	X,X
D5S818	NT	13,13	11,12
FGA	21,22,23	22,25	21,21

NT- not tested

Ignoring loci with “missing” alleles

- Some labs claim that this is a “conservative” statistic
- Ignores potentially exculpatory information
- “It fails to acknowledge that choosing the omitted loci is suspect-centric and therefore prejudicial against the suspect.”
  - Gill, et al. “DNA commission of the International Society of Forensic Genetics: Recommendations on the interpretation of mixtures.” FSI. 2006.

Mixtures where allelic drop out may have occurred

- Determining the rate of allelic drop out is problematic
- Interpreting evidence in a way that is least favorable to a defendant is taboo

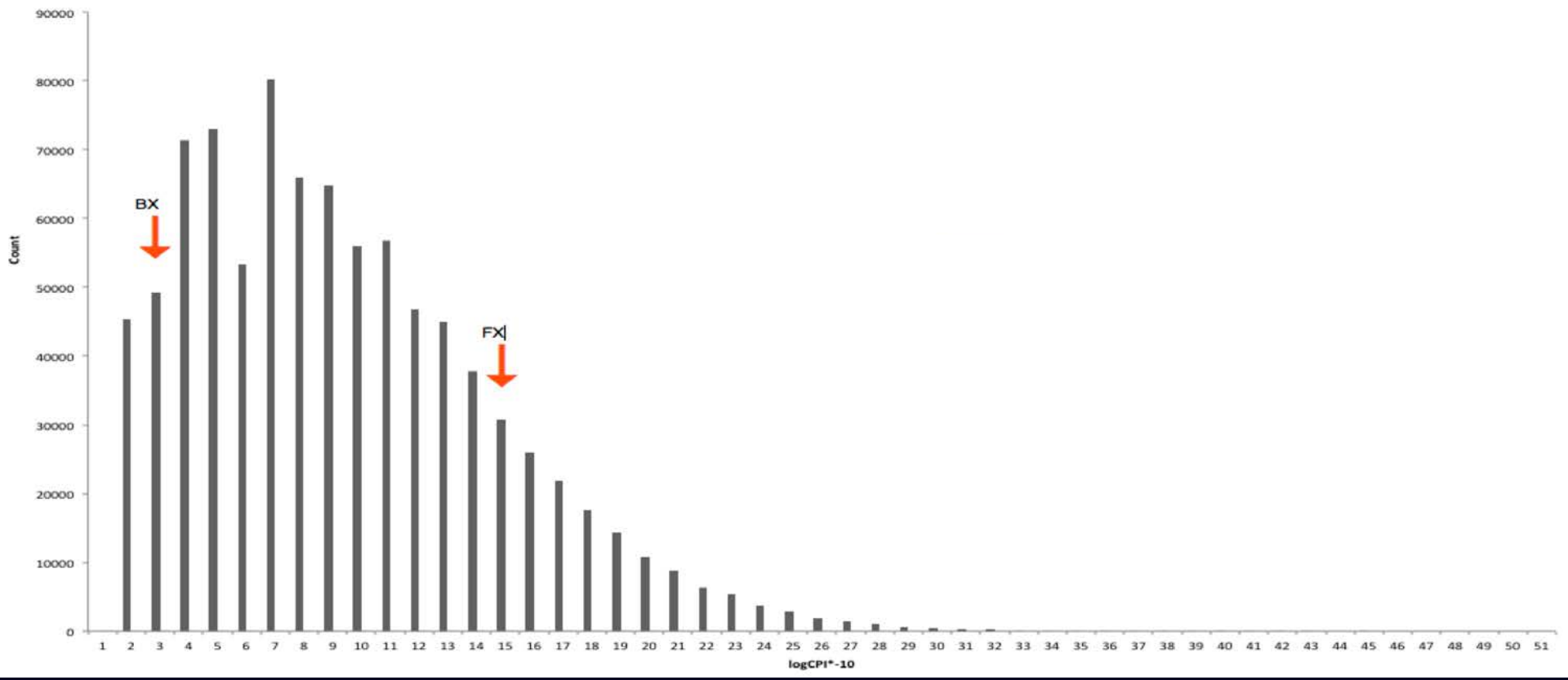
Mixtures where allelic drop out may have occurred

- Determining the rate of allelic drop out is problematic
- Interpreting evidence in a way that is least favorable to a defendant is taboo

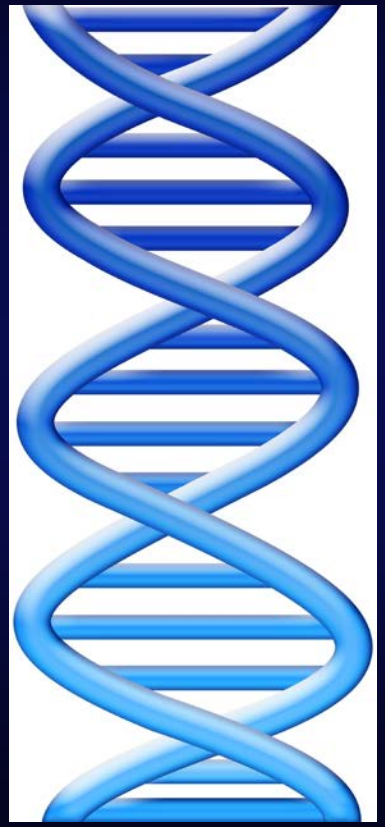
- What if instead of trying to assess drop out we invoke it liberally?
- What if instead of trying to eliminate knowledge of a reference profile we embrace it?

Mixtures with drop out: Combined Probability of Partial Inclusion (CPPI)

Combined Probability of Partial Inclusion



Mixed sample DNA statistics



- Weights for mixed samples are less than those for single source samples
- Determining the number of contributors can be difficult
- CPI can provide a statistic when no drop out has occurred

Statistical weights of mixed DNA profiles

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